ADOPT BBMRI-ERIC
gateway for health

ADOPT BBMRI-ERIC
GRANT AGREEMENT NO. 676550

DELIVERABLE REPORT

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BEST PRACTICE DOCUMENT FOR OPTIMAL USAGE OF OMICS TECHNOLOGIES FOR BIOMARKERS

Executive Summary

Different omics technologies are expected to accelerate biomarker discovery and validation but translation of these technologies into clinically actionable tools has been slow. Multiple technical platforms exist for omics profiling and there was a need to survey the operational and quality aspects of the omics technology platforms. With the help of the National Nodes, we produced a survey of the omics technology platforms in BBMRI-ERIC Member States. We received information from a total of 54 omics service providers from twelve BBMRI-ERIC Member States. Most of the results were obtained from genomics or transcriptomics platforms where the used technologies are already well validated. In the areas of metabolomics and proteomics the validation of technologies is still ongoing. The results of the questionnaires indicated that most of the omics service platforms are operating under quality control procedures but the quality of samples delivered by biobanks or researchers was generally not followed. The BBMRI-ERIC associated biobanks could clearly take a prominent position in jointly implementing international ISO and CEN/TS standards applicable for samples intended for omics analyses. Further collaboration with the technology platforms should be encouraged. The current results of the omics survey can support the biobankers and researchers to find omics technology platforms that have open access, operate under defined quality conditions and have

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been utilized to analyze high quality samples of BBMRI-ERIC biobanks or researchers utilizing biobank samples.

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1. Background

Omics technologies such as genomics, transcriptomics, metabolomics and proteomics enable simultaneous and holistic measurement of genes, gene expression, proteins and metabolites and these technologies are expected to accelerate the rate of biomarker discovery and validation. The promise of omics technologies is considered huge, but translation of these technologies into clinically actionable tools has been quite slow. Multiple technical platforms from different manufacturers are available for omics profiling and it is not always clear to the user which technology platform would give reliable results. Also, for biobanks it is essential to know those technology platforms that are able to return quality defined analysis data back to biobanks.

The fact is that standardized and harmonized protocols are still lacking within the omics field. Validation of omics technologies is far from complete and best practices for performing omics studies are yet to be established. Biobanks can participate in the development of best practices for omics studies because defined preanalytical conditions are a prerequisite for reproducible results. There is an increasing awareness of the need for high quality biospecimens and implementing QC tools assuring reliable results in biomarker studies.

More information of required sample quality and also quality of the technology providers is needed to reach the full potential of utilization of biobank samples in omics technologies. In this task we collaborated with the BBMRI-ERIC National Nodes to obtain information of omics technology platforms in BBMRI-ERIC Member States. The aim was to establish initial quality criteria and best practices for omics specific sampling for genomics, transcriptomics, metabolomics and proteomics.

With the help of the National Nodes, we produced a survey of the omics technology platforms in BBMRI-ERIC Member States. Furthermore, we collected here complementing information from the BBMRI-LPC project and other BBMRI initiatives.

2. Approaches (Methods)

The planning of the task was initiated in autumn of 2016. The topics for the questionnaires were discussed at the BBMRI-ERIC Management Committee (MC) meeting and a core working group, consisting of National Node directors from BBMRI.fi, BBMRI.at, BBMRI.nl and BBMRI.it was agreed and the omics questionnaire was planned in collaboration with ADOPT BBMRI-ERIC WP2. The questionnaire was created during May and June 2017. This included teleconferences between the core working group and consulting of 6 Finnish experts in omics analyses and technology platforms. The completed questionnaire was sent to the directors of BBMRI National Nodes in July 2017 with a cover letter, where they were asked to circulate the questionnaire to all relevant technology platforms in their country. Also, the BARC-database produced by BBMRI.se and BBMRI-LPC[1] was used to identify some of the relevant actors, which were contacted directly. The collection of data proceeded relatively slowly and the results of the questionnaire were presented and further discussed in the ADOPT project meeting of (Wroclaw,

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October 10th 2017). After the Wroclaw meeting the National Node directors took one more effort to get more answers to the questionnaires and we actually could almost double the amount of data. The answers to the questionnaires were received between July and early November and analyzed in November 2017.

3. Results

In total we received 54 answers to the 4 questionnaires. We received

- 23 answers to the genomics-questionnaire,
- 14 to the transcriptomics-questionnaire,
- 6 answers to the metabolomics-questionnaire
- 11 answers to the proteomics-questionnaire,

from omics technology platforms located in twelve BBMRI-ERIC Member States:

- Austria
- Estonia
- Finland
- France
- Germany
- Italy
- Latvia
- Malta
- Poland
- Sweden
- The Netherlands
- Turkey

The data collected in the questionnaires can be found in Appendix 2 a-d.

3.1 Genomics

The majority (16/23) of the institutions answering genomics-questionnaire reported to have open access. Majority of the institutions have determined required quality standards for the samples and the facility operates under strict quality control procedures. The most common quality standards for the facilities were ISO17025 and ISO9001, but also others were used (i.e; in France the French national norm for technological platform in research NFX 50-900). The quality standards required from the samples were more scattered and this is clearly an area where BBMRI-ERIC biobanks could have an impact. The quantity of required DNA varied from 50ng to 4µg. The required DNA concentrations varied between 2ng/µl to 60ng/µl, and most frequently 50ng/µl was required. Almost every facility was performing quality control to the samples, either themselves or through a close collaborator and almost everyone used Illumina as at least one of their technologies. Also raw files were delivered by the majority of the facilities, with other deliverables varying from comprehensive to none. It seems that the analysis technologies are well established but there is a need to agree on preanalytical standards of DNA samples and

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also the analysis and standardization of the results currently needs major input from the scientists of the receiving institute.

3.2 Transcriptomics

Most of the institutions answering the transcriptomics-questionnaire also reported to have open access (11/14). 7/14 respondents reported to have a quality standard for their facilities. Only one of the facilities had a ISO standard for the samples, while others required for example the RNA Integrity (RIN)- value to fall between 5-10. The quantity of required RNA needed for IVT reactions ranged from 1ng up to 1,5µg. The required concentrations varied between 10ng/µl to 500ng/µl. All facilities but one were performing quality control for their operations.

3.3 Metabolomics

Metabolomics services are currently produced only by a few technology platforms. We obtained data only from six platforms and two of them reported to have open access and being certified either for ISO 13485 or ISO 17025. One of the facilities was using NMR while others were doing MS-metabolomics. The required quantity of the serum/plasma was between 5-500µl. Every facility was doing quality control and the NMR platform has also initiated validation of their methodology with a clear aim in producing clinically relevant analysis tools.

3.4 Proteomics

We obtained data from 11 platforms and seven of them reported to have open access. Six of them had an official quality standard for the facility, all of which were using at least ISO9001. Only one of the facilities reported not to provide the quality control. The used technologies and throughput of deliverables varied largely between the facilities.

3.5 Omics related information collected in other initiatives

- BARC-database produced by BBMRI.se and BBMRI-LPC[1]
- BBMRI.nl has collected information from omics data collected in international collaboration activities. This data is found in the BBMRI-OMICS database[2]. It consists of omics data that has been generated and that is made available for BBMRI researchers focusing on integrative omics studies in Dutch Biobanks
- Core Technology for the Life Science network has technology platforms as members and more information on the omics technology platforms can be searched through the network web pages[3].

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3.6 BBMRI-ERIC operates actively towards biobank quality procedures

- Implementation of international ISO and CEN/TS applicable for biobanks[4]
- BBMRI-ERIC will organize a self-evaluation of biobanks during 2017[5]
- The ADOPT BBMRI-ERIC IT tool and Self-Assessment Survey[6] (D6.2) supports the structured monitoring of the pre-analytical process of samples as defined in the CEN/TS. This tool also provides key data for monitoring interoperability and quality of biobanks and appropriate collections.

4. Discussion and Conclusions

4.1 Towards best practices for optimal usage of omics technologies

The original aim of this survey was to produce a best practice document for the utilization of omics technologies for biobanks and researchers. The collection of initial information from the omics platforms was slow and as a result, a deeper analysis towards best practices could not be delivered within the ADOPT.

The current results of the omics survey can support the biobankers and researchers to find omics technology platforms that have open access, operate under defined quality conditions and have been utilized to analyze high quality samples of BBMRI-ERIC biobanks or researchers utilizing biobank samples.

Requirements for sample processing

- Biobanks should widely implement international ISO and CEN/TS standards applicable for samples to be delivered for omics analyses[5]
- Minimum requirement for the standard monitoring of pre-analytical processes of samples is advised to be monitored by using the ADOPT BBMRI-ERIC IT tool and Self-Assessment Survey[6,7]

Minimal requirements for the omics technology platform

- Technology platforms with open access should be preferred
- Technology platform should implement ISO quality standards for the platform
- A well-established technology platform performs a quality check of the imported samples. As a result, a double quality check for the samples is preferred

Test validation and documentation

- The omics technology platform should document all methods and procedures

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- Validation of method should be preferred

5. Next Steps

Discussions have been initiated towards collaboration with the BARC-database and BBMRI-LPC project participants to explore the possibilities to incorporate collected information with BARC-database. It would be important to identify a BBMRI-ERIC National node that would have interest in hosting the database of the omics technology platforms.

Close contacts and continuous discussion between biobanks and technology platforms will be necessary to further develop the awareness of the sample quality issues and to jointly develop the analytics. The coordination of such activities remains to be organized between BBMRI-ERIC headquarters and the National Nodes.

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6. References

[1]BARC-database: www.barcdb.org

[2]BBMRI-OMICS database: www.health-ri.org/services

[3]Core technology for the Life Science (CTLS) network: www.ctls-org.eu

[4]European Committee for Standardisation - cen.eu

[5]BBMRI-ERIC Quality Policy - www.bbmri-eric.eu/services/standardisation/

[6]BBMRI-ERIC Quality Management - www.bbmri-eric.eu/BBMRI-ERIC/quality-management/

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7. Appendices

Appendix I: List of omics technology platforms
- a) Based on genomics-questionnaire
- b) Based on transcriptomics-questionnaire
- c) Based on metabolomics-questionnaire
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Appendix II: a) Table of results for the genomics-questionnaire
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Appendix III: Abbreviations

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- a) Genomics-questionnaire
- b) Transcriptomics-questionnaire
- c) Metabolomics-questionnaire
- d) Proteomics-questionnaire

Appendix I: List of omics technology platforms

a) Based on genomics-questionnaire

Name of your facility	Country	Name	E-mail	Phone number
The Estonian Genome Center Core Facility	Estonia	Eneli Oitmaa	eneli.oitmaa@ut.ee	+3727375059
Helmholtz Zentrum München - GAC	Germany	Melanie Waldenberger	waldenberger@helmholtz-muenchen.de	+49 89 3187 1270
Biobank Lab, University of Lodz	Polska	Dominik Strapagiel	dominik.strapagiel@biol.uni.lodz.pl	+48693557721
FIMM genotyping unit	Finland	Kati Donner	kati.donner@helsinki.fi	+358 50 318 5677
The SNP&SEQ Technology Platform	Sweden	Tomas Axelsson	tomas.axelsson@medsci.uu.se	+46(0)701679458
Genome Database of Latvian population	Latvia	Davids Fridmanis	davids.fridmanis@biomed.lu.lv	+371 267808200

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Bioinformatics Long-term Support	Sweden	Björn Nystedt	bjorn.nystedt@scilifelab.se	018-471 4413
Clinical Genomics Uppsala	Sweden	Malin Melin	malin.melin@scilifelab.uu.se	+46184714656
Clinical Genomics Stockholm (SciLifeLab)	Sweden	Valtteri Wirta	valtteri.wirta@scilifelab.se	+46733386341
INT Biobank	Italy	Maria Grazia Daidone	mariagrazia.daidone@istitutotumori.mi.it	+390223902238
Leiden Genome Technology Center (LGTC)	The Netherlands	Susan Kloet	S.L.Kloet@lumc.nl	+31 71 5269441
Ankara University Biotechnology Institute	Turkey	Hilal Ozdag	hozdag@ankara.edu.tr	+905333717401
GenomeScan BV	Netherlands	Kees van den Berg	k.vandenberg@genomescan.nl	+31715681050
IBVR, IZSLER Biobank of veterinary resources	Italy	Stefano Pongolini	stefano.pongolini@izsler.it	+33-0521-293733
BaseClear bv.	The Netherlands	Björn te Boekhorst	info@baseclear.com	+31 (0)71 523 34 144
HUNT Biobank	Norway	Kristian Hveem	kristian.hveem@ntnu.no	+4747652530
Microsynth Austria	Austria	Lukas Hartl	lukas.hartl@microsynth.at	004369917246010
INRA Gentyane	France	Charles Poncet	charles.poncet@inra.fr	+33 443761519
FLUEXGEN	France	Nadège Brunel	nadege.brunel@inserm.fr	+33 14011441
NTNU Genomics Core Facility	Norway	Vidar Beisvåg	vidar.beisvag@ntnu.no	+47 728 25345
Translational Research	France	Abdel Bendahmane	abdel.bendahmane@inra.fr	+33 33169157687
Ligan-MP	France	Véronique Dhennin	veronique.dhennin@cnrs.fr	+33 374008127
MAD: Dutch Genomics Service & Support Provider	The Netherlands	Dr. Timo Breit	t.m.breit@uva.nl	+31-20-5257058

b) Based on transcriptomics-questionnaire

Name of your facility	**Country**	**Name**	**E-mail**	**Phone number**

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Estonian Genome Center Core Facility	Estonia	Eneli Oitmaa	eneli.oitmaa@ut.ee	7375059
Genome Database of Latvian population	Latvia	Davids Fridmanis	davids.fridmanis@biomed.lu.lv	+371 67808200
INT Biobank	Italy	Maria Grazia Daidone	mariagrazia.daidone@istitutotumori.mi.it	+390223902238
Ankara University Biotechnology Institute	Turkey	Hilal Ozdag	hozdag@ankara.edu.tr	+905333717401
SCIBLU Genomics	Sweden	Ingrid M. Rading	ingrid.magnusson_rading@immun.lth.se	0707185165
Microsynth Austria	Austria	Lukas Hartl	lukas.hartl@microsynth.at	004369917246010
IBENS Genomics facility	France	Stéphane Le Com	lecrom@biologie.ens.fr	NA
GenomEast Platform	France	Christelle Thibault-Carpentier	thibault@igbmc.fr	+33 388653426
Genomic Paris Centre IBENS	France	Corinne Blugeon	blugeon@biologie.ens.fr	+33 144322381
UCAGenomiX	France	UCAGenomiX	plateforme@ipmc.cnrs.fr	+33 493957790
NTNU Genomics Core Facility	Norway	Vidar Beisvåg	vidar.beisvag@ntnu.no	+47 728 25345
Ligan-MP	France	Véronique Dhennin	veronique.dhennin@cnrs.fr	+33 374008127
MAD: Dutch Genomics Service & Support Provider	The Netherlands	Timo Breit	t.m.breit@uva.nl	+31-20-5257058
Exploration of Metabolism platform, transcriptomic team	France	Céline Boby	celine.boby@inra.fr	+33 473624210

c) Based on metabolomics-questionnaire

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Name of your facility	Country	Name	E-mail	Phone number
Helmholtz Zentrum München, Genome Analysis Center	Germany	Jerzy Adamski	adamski@helmholtz-muenchen.de	+498931873155
Metanomics Health GmbH	Germany	Patrick Meikofski	patrick.meikofski@metanomics-health.de	+49 30 34807-410
Metabolomics Unit, FIMM, HiLIFE, UH.	Finland	Vidya Velagapudi	vidya.velagapudi@helsinki.fi	+358503175087
Nightingale Health Ltd	Finland	Karina Avetisyan	karina.avetisyan@nightingalehealth.com	+358407707517
ICANalytics	France	Farid Ichou	f.ichou@ican-institute.org	+33 142165238
Biomedical Metabolomics Facility Leiden	Netherlands	Thomas Hankemeier	hankemeier@lacdr.leidenuniv.nl	+31 71 527 4226

d) Based on proteomics-questionnaire

Name of your facility	Country	Name	E-mail	Phone number
Meilahti Clinical Proteomics Core Unit	Finland	Marc Baumann	marc.baumann@helsinki.fi	0294125200
Clinical Proteomics Platform	France	Christophe Hirtz	christophe.hirtz@umontpellier.fr	+33 603422349
Proteomics@PSL (SMBP-ESPCI)	France	Joëlle Vinh	joelle.vinh@espci.fr	+33 140795178
PP2I	France	Christian Larroque	christian.larroque@inserm.fr	+33 467618536
Functional Proteomics Platform	France	Philippe Marin	philippe.marin@igf.cnrs.fr	+33 434359213
PISSARO	France	Thierry Jouenne	thierry.jouenne@univ-rouen.fr	+33 235146680
Center for Proteomics and Metabolomics at the Leiden University Medical Center	Netherlands	Prof. Manfred Wuhrer	m.wuhrer@lumc.nl	+31-71-5268744
Protim	France	Charles Pineau	charles.pineau@inserm.fr	+33 223235279
Plateforme P3S - UMS omique	France	Olivier Silvie	olivier.silvie@inserm.fr	+33 140778132
OncoProteomics Laboratory	Netherlands	Connie Jimenez	c.jimenez@vumc.nl	020-4442340

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Centre for Molecular Medicine and Biobanking	Malta	Byron Baron	byron.baron@um.edu.mt	00356 79431224

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Appendix II: Tables of results

a) Table of results for the genomics-questionnaire

Name of facility	Country	Facility has open access	Quality standard of facility	Quality standard of samples	Quantity of DNA required	Concentration of DNA required	QC provided by the facility	Used Technology	Throughput capacity	Raw files inc. IDAT files	Genome Studio final report	.ped and .map files	zCall files	CNV files
The Estonian Genome Center Core Facility	EE	Yes	ISO 9000: 2008	ISO 9000:2008	up tp 4 ug	50 ng/ul	Yes	Illumina	2000-3000 per month	x				
Helmholtz Zentrum München - GAC	DE	Yes	ISO	non-degraded genomic DNA	500 ng genomic DNA	60 ng / µl	Yes	Affymetrix, Illumina, Agena	5000 samples / month	x	x	x		
Biobank Lab, University of Lodz	PL	Yes	In progress	Some IBBL PT	Depends on technique	Depends on platform - for arrays 50ng/mL	Yes	Illumina	800 ma/week, up to 200 smal genomes	x	x	x		x
FIMM genotyping unit	FI	Yes	None	Depends on who is sending us the samples	Yes	Depends on the technology	No	Affymetrix, Illumina, Agena	Depends on which technology is used	x	x	x	x	x
The SNP&SEQ Technology Platform	SE	Yes	ISO17025: 2005	ISO17025: 2005	200-400ng	min.15ng/µl, recommended 50ng/µl	Yes	Illumina	35K smpls/yr for SNP genotyping	x	x	x		x

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Genome Database of Latvian population	LV	Yes	none at the moment	Compliant with CEN/TS 16835-2	500 ng	over 50 ng/ul	Yes	Ion Torrent, Ion Proton	110 runs per year	x		x		x
Bioinformatics Long-term Support	SE	Yes	None	NA	NA	NA	No	Knoe-how in bioinfo omics (only staff)	25 large project per year					
Clinical Genomics Uppsala	SE	Yes	Some diagnostic tests are ISO15189	Genomics Facility (not biobank).	Highly dependent on method	Highly dependent on method	Yes	Illumina, NGS using several technologies,						
Clinical Genomics Stockholm (SciLifeLab)	SE	No	ISO 17025	-	variable	variable	Yes	Illumina	10000 samples					
INT Biobank	IT	only for colla-borative studies	ISO9001	not clear for me this query	50-500 ng	2-50 ng/microl	Yes	Affymetrix, Illumina, Agilent, Thermo Fisher	650/year	x	x			x
Leiden Genome Technology Center (LGTC)	NL	Yes	N/A	N/A	Depends on experiment	Depends on experiment	Yes	Illumina, PacBio, Bionano		x				
Ankara University Biotechnology Institute	TR	Yes	GLP is followed	DNA checked by gel electrophoresisOD260/OD280=1.75-1.85	312.5 ng per 250K SNP array	50 ng/ul	Yes	Affymetrix	30-50/month	x				x
GenomeScan BV	NL	NO	ISO 17025/GLP	various	various	various	Yes	Affymetrix. Illumina, PacBio/ LifeTech						

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IBVR, IZSLER Biobank of veterinary resources	IT	no	ISO 17025	purified dsDNA	300 ng	up to 10 ng/ul	Yes	Illumina	200 samples/ year	x				
BaseClear bv.	NL	yes	ISO 17025	?	We can work with very low quantities	We can work with very low quantities	Yes	Illumina, PacBio; Oxford Nanopore	Hundreds per week			x	x	x
HUNT Biobank	NO	Yes	ISO 9001	ISO 9001	200 ng	25-50 ng/ul	Yes	Illumina	4000 chiparrays/ week	x	x	x	x	x
Microsynth Austria	AT	no	ISO 9001: 2015,ISO/IEC 17025: 2005, GMP	we have no sample collection	0.5 µg	10 ng/µl	Yes	Illumina, Sanger Sequencing		x				
INRA Gentyane	FR	yes	ISO 9001: 2015: NF X 50-900	high	100 ng - 10µg	10 - 50 ng/µl	Yes	Affymetrix, Illumina, Pacific Bioscience	8 chips Affymetrix/ week, 14 runs Sequel PacBio/ week	x	x		x	x
FLUEXGEN	FR	no	ISO 9001	cDNA ozyme	3 µL	5ng/mL	Yes	fluidigm biomark HD		x				
NTNU Genomics Core Facility	NO	Yes	Illumina CSPro	-	-	-	Yes	Illumina		x	x	x		
Translational Research	FR	ips2.u-psud.fr	IBISA & CNOC (national standards)	Not relevant	Not relevant	Not relevant	Yes	Targeting Induced Local Lesions in Genomes	30 genes/ month					
Ligan-MP	FR	access via services & collab.	no	ratio 260/ 280: >1,8	500 ng	50ng/µl	No	Illumina	96 arrays / week	x	x	x	x	x

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MAD: Dutch Genomics Service & Support Provider	NL	Yes	Experience	Provided by customers	Depends on the application	Depends on the application	Yes	Affymetrix, NGS: Ion Proton and OxfordNano por	> 60 small to large microarray/ NGS projects	x		x	x	x

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b) Table of results for the transcriptomics-questionnaire

Name of facility	Country	Facility has open access	Quality standard of facility	Quality standard of samples	Quantity of RNA required	Concentration of RNA required	QC provided by the facility	Used technology	Throughput capacity	Deliverable 1	Deliverable 2	Deliverable 3	Deliverable 4
Estonian Genome Center Core Facility	EE	yes	ISO 9001:2008	ISO 9001:2008	minimum 0,5 ug	50 ng/ul	Yes	Illumina	2000	raw data			
Genome Database of Latvian population	LV	Yes	none at the moment	RIN nuber over 6	over 100 ng	10 ng/ul	Yes	Ion Torrent, Ion Proton	110 runs per year	Raw files	FASTQ files	Differentially expressed gene list	
INT Biobank	IT	only for collabo-rative studies	ISO9001	unclear query for me	50 ng- 1 µg	10 - 100 ng/ microl	Yes	Affymetrix, Illumina, ThermoFisher, Agilent, QuantStudio 12K Flex	2,500/year	BAN FASTQ files	CEL files	TXT files	
Ankara University Bio technology Institute	TR	Yes	GLP is followed though no certification for ISO etc.	RNA integrity by Bioanalyzer (min RIN 7.5), OD260/ OD280=1.9-2	1.5 ug for IVT reactions	500 ng/ul	Yes	Affymetrix	30-50/month	CEL	CHP	Differentially expressed gene list	Cluster, Pathway and Ontology analyses results
SCIBLU Genomics	SE	Yes	No	Pure RNA,RIN as close to 10 as possible.	100-200 ng	ca 100 ng/µl	Yes	Affymetrix	usually ca 1000 samples per year				
Microsynth Austria	AT	no	ISO 9001:2015, ISO/IEC 17025:2005	we have no sample collection	e1µg	e20ng/µl	Yes	Illumina	-	5 Mio reads, 1*75 or 1*150	10 Mio reads, 1*75 or 1*150	30 Mio reads, 1*75 or 1*150	50 Mio reads, 1*75 or 1*150
IBENS Genomics facility	FR	Yes	ISO 9001 NFX 50-900	Verified by the facility (qubit, bioanalyzer)	10 ng min.	200 ng/µL	Yes	Illumina	70 project and 800 samples / year	Fastq files	FastQC and FastqScreen reports	Read alignements and counts	Differential analysis

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GenomEast Platform	FR	yes	ISO9001, NFX50-900	NA	from 1 ng to 1 µg	Not required	Yes	Illumina	3500 samples per year	FASTQ files	md5 of FASTQ files	Samples and librairies quality control results	Sequencing quality control reports
Genomic Paris Centre IBENS	FR	yes	ISO 9001; NFX 50-900	more than 5 if possible	min 1 ng to begin	200 ng/µl if possible	Yes	Illumina, Oxford Nanopore Techno-logies	-	fastq and fastqc	filtered alignment files (BAM and BAI formats), files for genome viewers (bedGra	abundance measure-ment files (XLSX format)	matrix of the standard data (format XLSX), list of transcripts differentially ex
UCA GenomiX	FR	yes	ISO9001	very good	100 ng	1	Yes	Illumina, 10x Genomics	1Tb/month				
NTNU Genomics Core Facility	NO	Yes	Illumina CSPro	-	-	-	Yes	Illumina	-				
Ligan-MP	FR	access via services and collabo-rations	Not yet. We plan to work using ISO 15189	RINs for each sample by the customer	to be discussed	depending on each project	No	Illumina, RNA sequencing	We work with NextSeq and Hiseq4000	FastQ data	counts	other on demande	
MAD: Dutch Genomics Service & Support Provider	NL	yes	Experience	provided by customer	Depends on the application	Depends on the application	Yes	Affymetrix, IoProton-small-RNA-seq	>60 small & large projects per year	Anything a customer demands..	Lots of specialized bio-informatics		
platform "Exploration of Metabolism", transcriptomic team	FR	Yes	internal quality standard	MIQE	1 µg	25 ng/µl	Yes	Agilent microarrays - Applied Biosystems (q PCR)	-				

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c) Table of results for the metabolomics-questionnaire

Name of facility	Country	Facility has open access	Quality standard of facility	Quality standard of samples	Quantity of serum/ plasma required	QC provided by the facility	Used technology	Throughput capacity	Deliverable 1	Deliverable 2	Deliverable 3	Deliverable 4	Deliverable 5
Helmholtz Zentrum München, Genome Analysis Center	DE	no	research	research	depending on the assay (50-500 µL)	Yes	MS-metabolomics						
Metanomics Health GmbH	DE	No	Proprietary Work Instructions	SOPs	50µL per sample	Yes	MS-metabolomics		Sample type check	Blood Processing Control	Coagulation Quality Control	Sample Processing Control	Overall Quality
Metabolomics Unit, FIMM, HiLIFE, UH.	FI	Yes	Internal QC	Checked	100 µL	Yes	MS-metabolomics	2000 samples per year	Few weeks	Report in Word format	Data in Excel format	Results in ppt format	Manuscript writing
Nightingale Health Ltd	FI	No	EN ISO 13485	high	100µl	Yes	NMR	40000	228 metabolic biomarkers	Absolute quantified metabolites			
ICANalytics	FR	Yes	ISO 9001 (in progress)	French standard - NF S96-900 (in progress)	50-250 µl depends on LC-MS method	Yes	MS-metabolomics	4000 samples per year	Excel file - DataMATRIX	.pdf Report	.ppt and meeting		
Biomedical Metabolomics Facility Leiden	NL	users do not operate instruments	ISO 17-025	study pool is used as QC	from 5 µl to 500µl depending on target compounds	Yes	MS-metabolomics	15000 per year	validated targeted platforms	global profiling	miniaturised, 3D cell culture	lipidomics	

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d) Table of results for the proteomics-questionnaire

Name of facility	Country	Facility has open access	Quality standard of facility	Quality standard of samples	Quantity of serum/ plasma required	QC provided by the facility	Tech 1	Tech 2	Tech 3	Tech 4	Throughput capacity	Deliverable 1	Deliverable 2	Deliverable 3	Deliverable 4	Deliverable 5
Meilahti Clinical Proteomics Core Unit	FI		GLP	GLP	micrograms	Yes	clinical proteomics, MRM, SRM, PRM, Imaging MS	Glyco-proteomics, carbohydrate analysis and identification	systems proteomics, -medicine and -biology	DIGE, 1D and 2D analysis, micro and nano UHPLC	120 clin prot.P/M, 120 glyco prot.P/M, 200-400 IMS P/M,					
Clinical Proteomics Platform	FR	yes	ISO 9001	none	1 to 100 µL	Yes	Triple quadrupole MS	High Resolution MS	Digital ELISA	Multiplex ELISA	5000 samples/ year					
Proteomics @PSL (SMBP-ESPCI)	FR	yes	N/A	N/A	100µL	Yes	MALDI TOF/TOF	nanoESI FTICR	nanoESI Q Exactive (HF)	nanoESI LTQ Orbitrap	600-800/month in LCMS, more in MS only	identification of protein by peptide sequencing	stable isotope or label free quantification	redox state of proteins (oxidation, nitrosylation)	whole protein mass analysis	interactome and protein complexes study
PP2I	FR	CONTROLLED ACCESS ONLY	ISO9001	NO	Depending of the type of analyse	Yes	MALDI TOF	ESI TRIPLE TOF	CESI		60 TO 100 ANALYSES PER MONTH	feasability with our equipments	sensitivity required	sample processing	protein identification and/or quantification	
Functional Proteomics Platform	FR	YES	ISO9001-2015	Quidelines of high ranked journals	10 µg	Yes	nanoLC-FT-MS/MS				200/month	1-2 months delay				
PISSARO	FR	yes	ISO 9001, N-FX-50-900	none	few µL	Yes	mass spectrometry	Chromate-graphy	N-terminal micro-sequencing	Electro-phoresis	1000 samples/ month	protein identification	Post-translational modiifcations	N-terminal sequence	peptide sequencing	
Center for Proteomics and Metabolomics at the Leiden University Medical Center	NL	no	none	-80 and -20 sample storage, dependent technology applied	2 µl to 100 µl	Yes	Mass spectrometry glycomics of serum or plasma	NMR meta-bolomics of biofluids	targeted lipidomics platform by LC-MS		up to 2000 samples per month, depends on technology					
Protim	FR	yes	ISO 9001 v2015 & NF X50-900v2016	No	1 mL	Yes	shotgun proteomics	label free differential proteomics	post-translational modification analysis	mass spectrometry imaging	700 samples per year	N/A	N/A	N/A	N/A	N/A

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Plateforme P3S - UMS omique	FR	Yes	ISO9001	none	200 µL - Less to be tested	Yes	nanoLC-MS/MS ESI-Trap	MALDI-TOF/TOF	High resol nanoLC-MS/MS soon available		1000 / year					
OncoProteomics Laboratory	NL	yes	we comply with CCKL	N/A (mainly patient samples and validated cancer cell lines)	10 ml (platelet isolation)	Yes	nanoLC	tandem mass spectrometry			~4000-5000 nanoLC-MS/MS runs per year	turnaround time small-scale experiments 4-8 weeks	turnaround time large-scale experiments 3-6 months			
Centre for Molecular Medicine and Biobanking	MT	no	no	no	100µl-2ml	No	western blotting	ELISA				biomarker discovery	assay design and testing	population studies		

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Appendix III: Abbreviations

.map - A text file with no header file, and one line per variant with the following 3-4 fields: 1. Chromosome code 2.Variant identifier 3. Position in morgans/centimorgans 4.Base-pair coordinate

.ped – Original standard text format for sample pedigree information and genotype calls. Normally must be accompanied by a .map-file.

BAM – A binary format for storing sequence data

CEL – A data file created by Affymetrix DNA microarray image analysis software. Contains data extracted from probes on an Affymetrix GeneChip and can store thousands of data points.

CEN – European standardization organisation

CEN/TS – A Technical Specification is a normative document, used when various alternatives wouldn't gather enough to allow agreement on a EN, but need to coexist in anticipation for future harmonization or for providing specifications in experimental circumstances/evolving technologies

CHP – File format that contains probe set analysis results generated from Affymetrix software

CNV - Word data conversion support file

EN – Standard confirmed by European standardisation organisation CEN

FASTQ - A text-based format for storing both a nucleotide sequence and its corresponding quality scores.

GLP – Good Laboratory Practice

IBBL PT – Proficiency Testing programme provided by the Integrated BioBank of Luxembourg

IDAT – A file format used to store BeadArray data from the myriad of genome wide profiling platforms on offer from Illumina Inc.

ISO – International Standardization Organization

IVT – In Vitro Transcriptomics

MS – Mass Spectrometry

NGS – Next Generation Sequencing

OD – Optical Density

QC – Quality Control

RIN – RNA Integrity Number

SNP – Single Nucleotide Polymorphism

TXT – Filename extension for text files

WP – Work Package

zCall - A variant caller specifically designed for calling rare single-nucleotide polymorphisms from array-based technology

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Appendix IV: Questionnaires

a) Genomics-questionnaire

Omics technology platform questionnaire - Genomics

We are gathering information of the omics technology service providers in Europe. The information will be used to improve the biobanks' knowledge of possible omics technology service providers.

We hope you have time to answer this short questonnaire and we appreciate all your help and effort. This is a joint deliverable in ADOPT676550 -project (Horizon2020) Work Packages 2 and 6.

1. BACKGROUND INFORMATION

* Name:
* E-mail:
* Phone number:
* Name of your biobank/facility:
* Country:
* Does your biobank/facility have open access:

2. TECHNOLOGIES

Which technologies is your biobank/facility using?

- [ ] Affymetrix
- [ ] Illumina
- [ ] Other

If other, please specify:

Throughput capacity of your biobank/facility (per month or per year):

3. QUALITY

* Quality standard of your biobank/facility (ISO, CEN, etc.):
* Quality standard of the samples:

4. REQUIRED DNA

* Quantity of DNA required:
* Concentration of DNA required:

Quality control is provided by your biobank/facility:

- [ ] Yes
- [ ] No

Other comments:

5. DELIVERABLES

Deliverable throughput

- [ ] Raw files including IDAT files
- [ ] GenomeStudio final report
- [ ] .ped and .map files compatible with software such as Plink
- [ ] zCall files
- [ ] CNV files

PROCEED

Submit

End of the survey, thank you for your time!

This project has received funding from the *European Union's Horizon 2020 research and innovation programme* under grant agreement No 676550.

b) Transcriptomics-questionnaire

Omics technology platform questionnaire - Transcriptomics

We are gathering information of the omics technology service providers in Europe. The information will be used to improve the biobanks' knowledge of possible omics technology service providers.
We hope you have time to answer this short questonnaire and we appreciate all your help and effort. This is a joint deliverable in ADOPT676550 -project (Horizon2020) Work Packages 2 and 6.

1. BACKGROUND INFORMATION

* Name:
* E-mail:
* Phone number:
* Name of your biobank/facility:
* Country:
* Does your biobank/facility have open access:

2. TECHNOLOGIES

Which technologies is your biobank/facility using?

- [ ] Affymetrix
- [ ] Illumina
- [ ] Other

If other, please specify:
Throughput capacity of your biobank/facility (per month or per year):

3. QUALITY

* Quality standard of your biobank/facility (ISO, CEN, etc.):
* Quality standard of the samples:

4. REQUIRED RNA

* Quantity of RNA required:
* Concentration of RNA required:

Quality control is provided by your biobank/facility:

- [ ] Yes
- [ ] No

Other comments:

5. DELIVERABLES

Deliverable throughput 1:
Deliverable throughput 2:
Deliverable throughput 3:
Deliverable throughput 4:
Deliverable throughput 5:

PROCEED

Submit

End of the survey, thank you for your time!

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c) Metabolomics-questionnaire

Omics technology platform questionnaire - Metabolomics

We are gathering information of the omics technology service providers in Europe. The information will be used to improve the biobanks' knowledge of possible omics technology service providers.
We hope you have time to answer this short questonnaire and we appreciate all your help and effort. This is a joint deliverable in ADOPT676550 -project (Horizon2020) Work Packages 2 and 6.

1. BACKGROUND INFORMATION

* Name:
* E-mail:
* Phone number:
* Name of your biobank/facility:
* Country:
* Does your biobank/facility have open access:

2. TECHNOLOGIES

Which technologies is your biobank/facility using?

- [ ] NMR
- [ ] MS-METABOLOMICS
- [ ] Other

If other, please specify:
Throughput capacity of your biobank/facility (per month or per year):

3. QUALITY

* Quality standard of your biobank/facility (ISO, CEN, etc.):
* Quality standard of the samples:

4. REQUIRED SERUM/PLASMA

* Quantity of serum/plasma required:

Quality control is provided by your biobank/facility:

- [ ] Yes
- [ ] No

Other comments:

5. DELIVERABLES

Deliverable throughput 1:
Deliverable throughput 2:
Deliverable throughput 3:
Deliverable throughput 4:
Deliverable throughput 5:

PROCEED

Submit

End of the survey, thank you for your time!

This project has received funding from the *European Union's Horizon 2020 research and innovation programme* under grant agreement No 676550.

d) Proteomics-questionnaire

Omics technology platform questionnaire - Proteomics

We are gathering information of the omics technology service providers in Europe. The information will be used to improve the biobanks' knowledge of possible omics technology service providers.
We hope you have time to answer this short questonnaire and we appreciate all your help and effort. This is a joint deliverable in ADOPT676550 -project (Horizon2020) Work Packages 2 and 6.

1. BACKGROUND INFORMATION

* Name:
* E-mail:
* Phone number:
* Name of your biobank/facility:
* Country:
* Does your biobank/facility have open access:

2. TECHNOLOGIES

Which technologies is your biobank/facility using?

Technology 1:
Technology 2:
Technology 3:
Technology 4:
Throughput capacity of your biobank/facility (per month or per year):

3. QUALITY

* Quality standard of your biobank/facility (ISO, CEN, etc.):
* Quality standard of the samples:

4. REQUIRED SERUM/PLASMA

* Quantity of serum/plasma required:
Other sample types (cells, biopsies, tissues) - specify sample types and quantity required:

Quality control is provided by your biobank/facility:

- [ ] Yes
- [ ] No

Other comments:

5. DELIVERABLES

Deliverable throughput 1:
Deliverable throughput 2:
Deliverable throughput 3:
Deliverable throughput 4:
Deliverable throughput 5:

PROCEED

Submit

End of the survey, thank you for your time!

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